







## INTERNATIONAL APPLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

(51) International Patent Classification 7:
C12Q 1/00

A2

(11) International Publication Number: WO 00/50629

(43) International Publication Date: 31 August 2000 (31.08.00)

(21) International Application Number:

PCT/FI00/00140

(22) International Filing Date:

22 February 2000 (22,02,00)

(30) Priority Data:

990380

22 February 1999 (22,02,99) FI

(71) Applicant (for all designated States except US): ANCOBIO LTD. [GB/GB]; 375 West Regent Street, Glasgow G2 4LH (GB).

(72) Inventors; and

(75) Inventors/Applicants (for US only): JACOBS, Howard [GB/FI]; Näsilinnankatu 33 a D 14, FIN-33200 Tampere (FI). ROVIO, Anja [FI/FI]; Noormarkunkuja 4, FIN-36200 Kangasala (FI).

(74) Agent: KOLSTER OY AB; Iso Roobertinkatu 23, P.O. Box 148, FIN-00121 Helsinki (FI). (81) Designated States: AE, AL, AM, AT, AT (Utility model), AU, AZ, BA, BB, BG, BR, BY, CA, CH, CN, CR, CU, CZ, CZ (Utility model), DE, DE (Utility model), DK, DK (Utility model), DM, EE, EE (Utility model), ES, FI, FI (Utility model), GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KR (Utility model), KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, NO, NZ, PL, PT, RO, RU, SD, SE, SG, SI, SK, SK (Utility model), SL, TJ, TM, TR, TT, TZ, UA, UG, US, UZ, VN, YU, ZA, ZW, ARIPO patent (GH, GM, KE, LS, MW, SD, SL, SZ, TZ, UG, ZW), Eurasian patent (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European patent (AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE), OAPI patent (BF, BJ, CF, CG, CI, CM, GA, GN, GW, ML, MR, NE, SN, TD, TG).

## Published

Without international search report and to be republished upon receipt of that report.

(54) Title: DIAGNOSTIC AND SCREENING METHOD

(57) Abstract

The present invention relates to a novel diagnostic method for the detection of infertility in males. In particular, the present invention relates to a diagnostic method for detecting the presence or absence of a mutation or mutations in the *POLG* gene encoding mitochondrial DNA polymerase in a biological sample. The invention relates also to the use of a mutant *POLG* gene in the detection of infertility in males and in the screening of human populations for the presence of such mutation or mutations as a predictive test for male infertility. The invention also relates to the use of the *POLG* gene as an indicator of other pathological conditions associated with or related to male infertility, including those manifesting in women.

APPLICATION USE AS SPECIFICATION.